Correlations between mutations site in MEFV gene and phenotype of periodic disease, gout, palindromic rheumatisem and recurrent synovitis of hip

Abstract

Background and objective: *Auto-inflammatory syndromes have a distinct group in rheumatologic diseases. The aim of in this study, Correlations between mutations site in MEFVgene and phenotype of periodic disease, gout, palindromic rheumatisem and recurrent synovitis of hip.*

Methods: *This study is a case-control study. In this study, after a clinical examination and selection of patients: a) gout, b) palindromic syndrome and c) recurrent sinusitis, 32 patients were enrolled. All patients received complete history and demographic data, duration of symptoms, duration of diagnosis, type of disease, etc, and were obtained in checklist of each patient. Then blood samples were taken from the patients and taken to the Genetic Laboratory for examination of the varieties or mutations obtained for the MEFV gene. After determining the MEFV gene and its sequencing in patients, all of the data entered the SPSS v21 statistical program and finally we analyzed the data.*

Results: *In this study, the mean age of patients with gout was 53.41 years. The majority of patients were male and thumb was involved most commonly joint. It was also observed that 20.8% of patients had MEFV positive gene and the most common genotype E148Q in 16.7% of patients. Among 5 patients with palindromic syndrome, the mean age of patients was 14.6 years and Most of them were male and hip and knee were the most commonly involved joints. It was also observed that only 1 (20%) patients had MEFV gene and the V726A / Wt gene was reported. Of the 3 patients with Periodic Synovite, the mean age of the patients 15.66 years and most of them were male. It was also observed that all three patients with periodontal sinus had MEFV gene mutations including R761H / Wt, V726A / Wt and A744S / Wt.*

Conclusion: *In the present study, MEFV gene was observed in all patients with Periodic Synovite. Also, a high percentage of patients with gout and palindromic syndrome had positive gene mutations.*

Keywords: *MEFV Gene, Gout, Palindromic Rheumatism, Hip Recurrent Synovite*